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Peculiarities in the gestural repertoire: An early marker for Rett syndrome?

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ABSTRACT

We studied the gestures used by children with classic Rett syndrome (RTT) to provide evidence as to how this essential aspect of communicative functions develops. Seven participants with RTT were longitudinally observed between 9 and 18 months of life. The gestures used by these participants were transcribed and coded from a retrospective analysis of a video footage. Gestures were classified as deictic gestures, play schemes, and representational gestures. Results of the analysis showed that the majority of gestures observed were of deictic character. There were no gestures that could be classified as play schemes and only two (e.g., head nodding and waving bye bye) that were coded as representational or symbolic gestures. The overall repertoire of gestures, even though not necessarily delayed in it's onset, was characterized by little variability and a restricted pragmatic functionality. We conclude that the gestural abilities in girls with RTT appear to remain limited and do not constitute a compensatory mechanism for the verbal language modality.

1. Introduction

Rett syndrome (RTT) is a severe neurodevelopmental disorder that generally affects females and is mainly caused by mutations in the X-linked *MECP2* gene (Amir et al., 1999; Neul et al., 2010). It is associated with severe intellectual disability, autistic-like behavior, communicative restrictions and difficulties in hand use coinciding with specific stereotyped movements such as hand-wringing or washing-like movements (Carter et al., 2010; Cass et al., 2003; Hagberg, Aicardi, Dias, & Ramos, 1983; Kaufmann et al., 2012; Kerr, Archer, Evans, & Gibbon, 2006; Matson, Fodstad, & Boisjoli, 2008; Neul et al., 2010). One of the necessary criteria for classic RTT is a recognizable regression that is followed by a period of recovery or stabilization (Neul et al., 2010). Regression is defined by a loss of previously acquired skills, specifically spoken language and purposeful hand use. The general development before regression was initially believed to be asymptomatic, but recent

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research has provided mounting evidence of subtle abnormalities in the motor, speech-language and communicative repertoires during the first year of life (e.g. Burford, 2005; Einspieler, Kerr, & Prechtl, 2005a,b; Kerr et al., 2006; Leonard & Bower, 1998; Marschik, Einspieler, Oberle, Laccone, & Prechtl, 2009; Marschik, Einspieler, & Sigafoos, 2012; Marschik, Kaufmann, et al., in press; Marschik, Pini, et al., 2012; Tams-Little & Holdgrafer, 1996).

The recent description of early peculiarities in the speech-language and motor domain have contributed to a better understanding of how *MECP2* deficiencies influence early (functional) brain development, and consistent additions to this body of knowledge might lead to a more timely diagnosis. As diagnosis to date is usually made at a mean age of 3 years and usually after the onset of regression (Fehr et al., 2011; Laurvick et al., 2006), there are limited possibilities to study the pre-regressional development (Marschik & Einspieler, 2011). Retrospective video analysis and retrospective questionnaires (parental interviews) are currently the methods of choice to track down early developmental peculiarities in speech-language and communicative development during this period. Questionnaires have proven to be a valuable source of documenting various concurrent behaviors including speech-language functions in children with developmental disabilities (Charman, Drew, Baird, & Baird, 2003; Luyster, Lopez, & Lord, 2007). However, retrospective assessments of communicative functions must be interpreted more cautiously when considering the following factors affecting reliability: (a) long time lag between the interview/questionnaire and period of interest; (b) memory bias of parents with affected children; and (c) the lack of parental training in the observation of linguistic or cognitive skills (Einspieler, Widder, Holzer, & Kenner, 1988; Marschik & Einspieler, 2011; Marschik, Einspieler, Garzarolli, & Prechtl, 2007). Consequently, the best available method for obtaining a detailed description of various developmental domains in children with late-clinical-onset disorders is the retrospective video analysis of non-standardized home movies (Einspieler et al., 2005a; Maestro et al., 2001; Marschik & Einspieler, 2011; Ozonoff et al., 2011; Palomo, Belinchón, & Ozonoff, 2006; Saint-Georges et al., 2010).

Given the methodological restrictions of assessing developmental profiles during the pre-regression period in RTT, there are only a few studies that report on early speech-language and communicative functions. The scaffolding function of early language capacities that precede and predict later ones is the reason why it is of high relevance to focus on this critical period of development in girls with RTT.

Increased language comprehension abilities appear to be a route to productive language abilities, and communicative gestures appear to facilitate this transition (Capone & McGregor, 2004; Iverson & Goldin-Meadow, 2005; Volterra & Erting, 1990). For example, in the first year of life, typically developing children begin to communicate through vocalizations, eye gaze, and gestures to express their needs and desires (e.g. Bates, Camaioni, & Volterra, 1975; Karmiloff & Karmiloff-Smith, 2001; Stone, Ousley, Yoder, Hogan, & Hepburn, 1997; Trevarthen & Hubley, 1978). Gestural development usually begins with the use of deictic gestures or pre-linguistic gestures (or performatives; e.g. showing, giving, pointing, reaching out) around 10–12 months of age. Performatives are usually observed a month before the first words (Bates, Benigni, Bretherton, Camaioni, & Volterra, 1979; Bates et al., 1975; Goodwyn & Acredolo, 1993) and followed by play schemes where children depict an object in terms of its functions. Representational (sometimes referred to as symbolic) gestures usually emerge before the 25-word milestone (Acredolo & Goodwyn, 1988; Capone & McGregor, 2004), and can be differentiated from play schemes in that the referent is not manipulated (e.g., using the hand to symbolize a flying airplane) and they do not change with context. Representational gestures are often complementary to spoken words (Iverson, Capirci, & Caselli, 1994), and as gestures and vocalizations simultaneously appear they increase the saliency of communicative acts that facilitate interpretations by the caregiver. This in turn increases parental responsiveness to communication (Karmiloff & Karmiloff-Smith, 2001; Vallotton, 2009; Yoder, Warren, Kim, & Gazdag, 1994).

Only a few studies have so far dedicated their focus of interest to the development of gestures in RTT. Lavås, Slotte, Jochym-Nygren, Van Doorn, and Witt-Engerström (2006) reported that, among a cohort of 125 girls with RTT, 50% were able to use eye-pointing, index finger pointing, or orther gestures, without further specifying the latter. Pre-regressional gesture use was first described by Tams-Little and Holdgrafer (1996) by means of parent-completed questionnaire. They also focused on forms and functions of gestures and speculated about having discovered an early marker for RTT. Our own studies on females with the preserved speech variant (PSV) of RTT, a milder variant with relatively better speech-language abilities, revealed restricted repertoires of their socio-pragmatic functions and communicative gestures during the second year of life after a period with abnormal inspiratory vocalizations (i.e. proto-vowel or proto-consonant alternations produced on ingressive airstream; Marschik, Einspieler, et al., 2012; Marschik, Kaufmann, et al., in press; Marschik, Pini, et al., 2012).

Because our previous studies of individuals with PSV revealed a restricted repertoire of intentional gestures (Marschik et al., 2009; Marschik, Kaufmann, et al., in press) we were curious as to how this essential aspect of communicative functions develops in individuals with classic RTT. In order to obtain a better understanding of the development of communicative gestures in females with RTT, we designed the present study to address the following questions: (1) At what age do intentional gestures first occur? (2) What gestures can be observed during the first 18 months of life in individuals with RTT? (3) How complex is the gestural repertoire during this age period? and (4) How can gestures be categorized (deictic/proto-symbolic/representational)?

2. Methods

2.1. Participants

The present study focused on the acquisition and composition of the gestural repertoire of seven females with RTT who were longitudinally observed between 9 and 18 months of life. Four were from English speaking families (Cases 1, 2, 3, and 6)

and three were from German speaking families (Cases 4, 5, and 7). All females were singletons, born as a result of uneventful pregnancies and deliveries with birth weights, birth lengths, occipitofrontal circumferences, and Apgar scores in the normal range. All participants were *MECP2* mutation-positive. The study was approved by the relevant research ethics committees. All parents gave their informed consent, including consenting to the publication of the results.

2.2. Procedure

This study is based on the retrospective analysis of a comprehensive video footage from typical family routines (play situations, bathing, feeding, etc.) and special events (such as religious festivals, birthdays or family gatherings) when the participants were between 9 and 18 months of age. All videos had been made by the females' parents, who were not aware at that time that their daughters had RTT. The analyses are based on 21 h of total footage comprised of 358 separate clips. A research assistant, naive to the purpose of the study, checked the recordings for sufficient length and quality standards, copied the relevant video recordings, and prepared them for analysis (unifying the codecs and sampling the recordings across the age range).

2.3. Assessment of the gestural repertoire

From the retrospective video footage we analyzed the occurrence and use of intentional gestures for communicative purpose. All gestures were coded and transcribed in chronological order using the Noldus Observer-XT. Each transcript and coding (by TW) was rechecked by a second transcriber (CE, KBP, or PBM) against the audio–video files in order to ensure accuracy and consistency. In case of disagreement, the video-sequences in question were discussed within the team until agreement was achieved. The final transcriptions were analyzed to classify gestures using the Austrian-German adaptations of the MacArthur-Bates Communicative Development Inventories, a checklist to assess early socio-communicative functions, early gestures, vocabulary and grammar (ACDI; Marschik et al., 2007).

3. Results

The extent of the gestural repertoire was limited to between one and six different gestures per child. The complexity and the composition of the gestural repertoire are given in Table 1. The first intentional gestures (demonstrating an object, head nodding, and waving bye bye) were observed during the 9th month of age. These earliest gestures were exhibited by Cases 4 and 7. Cases 2 and 3 showed their first communicative gestures during their 10th month of age; Case 1 one month later; and Case 6 at 12 months of age. Case 5, however, showed no gestures during any of her videotapes. The appearance of the first three gestures (only applicable to Cases 3 and 4; see below for the complexity of the repertoire and Table 1) was observed within the same month of the first gesture; Case 4 acquired the first three intentional gestures during the 9th month of age, and Case 3 during the 10th month of age.

The overall composition of the gestural repertoire consisted of the following gestures: (a) demonstrating an object, (b) passing on an object, (c) index finger pointing, (d) extending arms, (e) head nodding, and (f) waving bye bye. These observed gestures refer to the following potential communicative functions or purposes: (a) attention to self, (b) requesting an object, (c) requesting an action, (d) answering, and (e) imitation.

The classification of gestures into deictic gestures, play schemes, and representational gestures revealed the following results: the majority of gestures observed (4 out of 6 overall observed gestures) were of deictic character. There were no gestures that could be classified as play schemes. Two gestures were of representational or symbolic character, namely, head nodding (exhibited by Cases 1, 3, 4, and 7) and waving bye bye (Case 4). Both symbolic forms were only observed in Case 4, the participant who had the earliest onset of deictic gestures and the most complex gestural repertoire that consisted of six different intentional gestures (Table 1). Cases 2 and 6 exclusively showed gestures of deictic character, Case 5 – as mentioned above – did not display any gestures.

Table 1
Gestural repertoire of seven females with RTT between 9 and 18 months of life.

	Demonstrating an object	Passing an object	Index finger pointing	Extended arms seeking comfort	Head nodding indicating yes	Waving indicating bye bye	Total number of gestures used
Case 1	●	□	□	□	●	□	2
Case 2	□	□	●	□	□	□	1
Case 3	●	●	□	●	●	□	4
Case 4	●	●	●	●	●	●	6
Case 5	□	□	□	□	□	□	0
Case 6	●	□	□	□	□	□	1
Case 7	●	□	□	□	●	□	2

(●) presence and (□) absence of gestures.

4. Discussion

Since the first suggestions of atypical development during the pre-regression period in individuals with RTT, there has been a mounting body of knowledge regarding early functional abnormalities in RTT. This study is a continuation of our previous work reporting on early speech-language dysfunctions in RTT and its preserved speech variant based on retrospective video analysis (Marschik et al., 2009; Marschik, Einspieler, Prechtl, Oberle, & Laccone, 2010; Marschik, Einspieler, et al., 2012; Marschik, Kaufmann, et al., in press; Marschik, Lanator, Freilinger, Prechtl, & Einspieler, 2011; Marschik, Pini, et al., 2012). It sheds new light on a developmental domain that has been reported to be atypical in RTT, as indicated by retrospective parental questionnaires (Kerr et al., 2006; Lavås et al., 2006; Tams-Little & Holdgrafer, 1996). We have to keep in mind, however, the limitations of both methods, retrospective questionnaires and retrospective video analysis. The latter has limited value in assessing quantitative aspects of speech-language capacities, as recordings usually do not cover an exhaustive set of acquired capacities (Marschik & Einspieler, 2011). On the other hand, video analysis allows for direct observation of early speech-language abilities that cannot be reliably assessed using retrospective parent report due to limitations such as potential memory bias and restricted knowledge about communicative and linguistic development (Einspieler et al., 1988; Luyster et al., 2007; Marschik et al., 2007).

Using retrospective parental questionnaires, Tams-Little and Holdgrafer (1996) revealed that gesture use as a precursor for linguistic development was delayed in girls with RTT. Based on this finding – and on reports about delayed gestural development in children with developmental disabilities such as autism spectrum disorder, late talkers, children with specific language impairment, and individuals with acquired brain lesions (Capone & McGregor, 2004; Charman et al., 2003; Hill, Bishop, & Nimmo-Smith, 1998; Sauer, Levine, & Goldin-Meadow, 2010; Thal & Tobias, 1992) – we expected a delayed onset of the first intentional gesture(s) in girls with RTT. Contrary to our expectations and previous findings, the onset of the first communicative gestures was not delayed in our – admittedly small – sample. The two girls in our study who acquired at least three gestures (Cases 3 and 4) acquired them within one month from the first gesture. On the other hand, early acquisition does not necessarily predict a greater complexity of the gestural repertoire compared to the individuals displaying a later onset of gestures. Case 7, for example, acquired her first gesture at an age of 9 months, but she acquired only one more gesture during the assessment period.

If we have a closer look beyond the appearance of the first gesture(s) and the development of the gestural repertoire, the picture changes dramatically. The overall repertoire of gestures for all individuals was characterized by little variability and a restricted pragmatic functionality. We observed only six different gestures in the entire corpus. This restricted repertoire is in line with previous observations in RTT (Tams-Little & Holdgrafer, 1996) and also in individuals with autism who were reported to have a limited repertoire of gestures and a lower proportion of gestures combined with vocalizations as compared to typically developing children (Landa, 2008; Wetherby, Yonclas, & Bryan, 1989). The pragmatic functions covered by the gestural repertoire (Table 1) are restricted to attention to self, requesting an object, requesting an action and imitation (Sigafoos, Arthur-Kelly, & Butterfield, 2006; Sigafoos, Woodyatt, Keen, et al., 2000). The repertoire, consisting of demonstrating and passing an object, finger pointing, extending arms toward the caregiver, nodding with the head, and waving bye bye is comparable to the repertoire reported in girls with PSV of RTT (Marschik, Kaufmann, et al., in press). In addition, as reported earlier (Dahlgren Sandberg, Ehlers, Hagberg, & Gillberg, 2000; Marschik et al., 2009; Marschik, Kaufmann, et al., in press; Tams-Little & Holdgrafer, 1996), the individual gestural repertoires were very limited with a maximum of six different gestures (range 0–6; Table 1). The limited repertoire of gestures might result from the fact that girls with RTT were reported to have difficulties in focusing their attention to relevant sources of information and exhibit limited behaviors indicative of an intention to communicate (Fabio, Antonietti, Castelli, & Marchetti, 2009; von Tetzchner, 1997; Woodyatt & Ozanne, 1992a, 1992b, 1993). This is closely related to joint attention behaviors that are considered to play a central role in identifying gestures with communicative intent and in social communication in general (Dahlgren Sandberg et al., 2000; von Tetzchner, 1997). Indeed, a reduced intention to communicate influences the social-reciprocal system in that children developing atypically tend to be more passive in conversational activities, and that adults in turn are less likely to interact with passive children compared to more active ones (Karmiloff & Karmiloff-Smith, 2001).

Classification of the types of observed gestures revealed that four of the six gestures were of deictic character. No gestures were with play scheme character, but there were two symbolic gestures. We agree with Charman et al. (2003) in describing the gestures as more likely to be instrumental or functional actions rather than symbolic gestures per se. Furthermore, the gestures classified as symbolic gestures here (nodding with the head and waving bye bye) have to be seen in the light of imitations with perseverative character and in close relation to the emergence of stereotypies. Therefore, caution should be taken when making interpretations as some of the observed gestures might mimick communicative behaviors while lacking actual communicative intent. The repertoire of representational gestures might be considered as restricted as Acredolo and Goodwyn (1988) reported a mean number of three to five representational gestures in infant toddler gestural repertoires.

Another interesting finding was that index finger pointing was only observed in two girls: in Case 4 who had a repertoire of six gestures and also Case 2 who displayed index finger pointing as her only gesture. Again, this finding was comparable to those of girls with PSV of RTT in that these children also had limited pointing abilities (Marschik, Kaufmann, et al., 2012). This is in line with reports on children with RTT and ASD who overall displayed less pointing, showing objects, and less joint attention as compared to typically developing children (Charman, 1998; Nomura & Segawa, 1990; Shumway & Wetherby, 2009; Stone et al., 1997).

Besides the methodological restrictions of video analysis (Marschik & Einspieler, 2011) one of the limitations of this study is the small sample size that limits generalizability of the findings. Furthermore, the actual range of the gestural repertoire might have been broader than the amount extracted from the video footage. Case 5, for example, is reported here as displaying no gestures, but her footage was the shortest, and some months were not covered sufficiently. Consequently, these findings should be interpreted with caution as this individual may have potentially exhibited gestures that simply were not recorded. Furthermore, retrospective video recordings are not standardized and several factors may vary substantially among recordings, potentially affecting the assessment of communicative behaviors. Variation was often seen in the duration of the recordings, communicative setting (high vs. low communicative settings), number of people involved in the video, etc. (Matson, Wilkins, & González, 2008). However, video analysis may enable observers to clearly distinguish between potential communicative acts and preintentional communicative forms, i.e. caregivers assigning meaning to the child's behaviors (Sigafoos, Woodyatt, Tucker, Roberts-Pennell, & Pittendreigh, 2000; Woodyatt & Ozanne, 1993). Another issue to be addressed is the onset of regression in RTT, which is not easily defined and able to be ascribed to a definite date but is rather a gradual process of decline over various developmental domains. Regression in classic RTT as well as in its preserved speech variant is characterized by, at least, the loss of hand skills and productive language (Neul et al., 2010). Nevertheless, non-verbal communicative functions and motor skills could also be affected. Most commonly, regression takes place between 12 and 18 months, but in a few cases even before 6 months of age or after 36 months (Charman et al., 2002). The exact onset of regression is, however, difficult to define and might have started in at least some of the individuals of our study by the end of the observation period.

Considering that gestures are a predictor for later language development and scaffold language and cognitive development (Bates et al., 1975; Rowe & Goldin-Meadow, 2009; Tomasello, Striano, & Rochat, 1999), our findings might contribute to the early detection of RTT. We are cautious not to label it as a contribution to early diagnosis as this requires further study, validation and consideration of other developmental domains. "Early diagnosis is a good development only if the diagnoses are reliable, have good predictive validity, and are useful in assisting better care and prognosis", as Matson, Wilkins, et al. rightly stated (2008, p. 76). Nonetheless, our findings in combination with recent attempts to identify behavioral patterns deviant from typical development may facilitate early detection in the near future.

5. Conclusions

This study characterized another parameter of the deviant atypical character of communicative development in girls with classic RTT: specifically, the deficit in communicative gestures. Just like individuals with Down syndrome, girls with RTT have difficulties in verbal abilities. However, children with Down syndrome have a dissociative profile with the advantage that gesture use often compensates for verbal insufficiencies (Caselli, Vicari, Longobardi, Lami, & Pizzoli, 1998; Zampini & D'Odorico, 2009), whereas gestural abilities in girls with RTT remain limited and do not constitute a compensatory mechanism for the verbal language modality.

The use of gestures is considered to be an early index of global communicative skills (Rowe & Goldin-Meadow, 2009; Rowe, Özçalışkan, & Goldin-Meadow, 2008) and certain genetic disorders might have specific atypical communicative developmental trajectories. There is, however, still the need for a more detailed understanding of this issue to address the question of whether the observed pattern is specific to RTT. Despite our conflicting findings regarding the first appearance of gestures, we agree with Tams-Little and Holdgrafer (1996) that a limited repertoire of gestures in conjunction with qualitative peculiarities in other speech-language domains might be characteristic for a severe neurodevelopmental disorder like RTT.

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References

Acredolo, L., & Goodwyn, S. (1988). Symbolic gesturing in normal infants. *Child Development, 59*, 450–466.

Amir, R. E., Van den Veyver, I. B., Wan, M., Tran, C. Q., Francke, U., & Zoghbi, H. Y. (1999). Rett syndrome is caused by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2. *Nature Genetics, 23*, 185–188.

Bates, E., Benigni, L., Bretherton, I., Camaioni, L., & Volterra, V. (1979). Cognition and communication from nine to thirteen months: Correlational findings. In E. Bates (Ed.), *The emergence of symbols: Cognition and communication in infancy* (pp. 69–140). New York: Academic Press.

Bates, E., Camaioni, L., & Volterra, V. (1975). The acquisition of performatives prior to speech. *Merrill Palmer Quarterly, 21*, 205–226.

Burford, B. (2005). Perturbations in the development of infants with Rett disorder and the implications for early development. *Brain and Development, 27*, S3–S7.

Capone, N. C., & McGregor, K. K. (2004). Gesture development: A review for clinical and research practices. *Journal of Speech, Language, and Hearing Research, 47*, 173–186.

Carter, P., Downs, J., Bebbington, A., Williams, S., Jacoby, P., Kaufmann, W. E., et al. (2010). Stereotypical hand movements in 144 subjects with Rett syndrome from the population-based Australian database. *Movement Disorders, 25*, 282–288.

Caselli, M. C., Vicari, S., Longobardi, E., Lami, L., & Pizzoli, C. (1998). Gestures and words in early development of children with Down syndrome. *Journal of Speech, Language, and Hearing Research, 41*, 1125–1135.

Cass, H., Reilly, S., Owen, L., Wisbeach, A., Weekes, L., Slonims, V., et al. (2003). Findings from a multidisciplinary clinical case series of females with Rett syndrome. *Developmental Medicine and Child Neurology, 45*, 325–337.
Charman, T. (1998). Specifying the nature and course of the joint attention impairment in autism in the preschool years: Implications for diagnosis and intervention. *Autism, 2*, 61–79.
Charman, T., Cass, H., Owen, L., Wigram, T., Slonims, V., Weeks, L., et al. (2002). Regression in individuals with Rett syndrome. *Brain and Development, 24*, 281–283.
Charman, T., Drew, A., Baird, C., & Baird, G. (2003). Measuring early language development in preschool children with autism spectrum disorder using the MacArthur Communicative Development Inventory (infant form). *Journal of Child Language, 30*, 213–236.
Dahlgren Sandberg, A., Ehlers, S., Hagberg, B., & Gillberg, C. (2000). The Rett syndrome complex: Communicative functions in relation to developmental level and autistic features. *Autism, 4*, 249–267.
Einspieler, C., Kerr, A. M., & Prechtl, H. F. (2005a). Abnormal general movements in girls with Rett disorder: The first four months of life. *Brain and Development, 27*, 8–13.
Einspieler, C., Kerr, A. M., & Prechtl, H. F. (2005b). Is the early development of girls with Rett disorder really normal? *Pediatric Research, 57*, 696–700.
Einspieler, C., Widder, J., Holzer, A., & Kenner, T. (1988). The predictive value of behavioural risk factors for sudden infant death. *Early Human Development, 18*, 101–109.
Fabio, R. A., Antonietti, A., Castelli, I., & Marchetti, A. (2009). Attention and communication in Rett syndrome. *Research in Autism Spectrum Disorders, 3*, 329–335.
Fehr, S., Bebbington, A., Nassar, N., Downs, J., Ronen, G. M., De Klerk, N., et al. (2011). Trends in the diagnosis of Rett syndrome in Australia. *Pediatric Research, 70*, 313–319.
Goodwyn, S., & Acredolo, L. (1993). Symbolic gesture versus word: Is there a modality advantage for onset of symbol use? *Child Development, 64*, 688–701.
Hagberg, B., Aicardi, J., Dias, K., & Ramos, O. (1983). A progressive syndrome of autism, dementia, and loss of purposeful hand use in girls: Rett's syndrome: Report of 35 cases. *Annals of Neurology, 14*, 471–479.
Hill, E. L., Bishop, D. V. M., & Nimmo-Smith, I. (1998). Representational gestures in developmental coordination disorder and specific language impairment: Error-types and the reliability of ratings. *Human Movement Sciences, 17*, 655–678.
Iverson, J., Capirci, O., & Caselli, M. C. (1994). From communication to language in two modalities. *Cognitive Development, 9*, 23–43.
Iverson, J., & Goldin-Meadow, S. (2005). Gesture paves the way for language development. *Psychological Science, 16*, 367–371.
Karmiloff, K., & Karmiloff-Smith, A. (2001). *Pathways to language: From fetus to adolescent*. Cambridge. MA Harvard University Press.
Kaufmann, W. E., Tierney, E., Rohde, C. A., Suarez-Pedraza, M. C., Clarke, M. A., Salorio, C. F., et al. (2012). Social impairments in Rett syndrome: Characteristics and relationship with clinical severity. *Journal of Intellectual Disability Research, 56*, 233–247.
Kerr, A. M., Archer, H. L., Evans, J. C., & Gibbon, F. (2006). People with mutation positive Rett disorder who converse. *Journal of Intellectual Disability Research, 50*, 386–394.
Landa, R. J. (2008). Diagnosis of autism spectrum disorders in the first 3 years of life. *Nature Clinical Practice Neurology, 4*, 138–147.
Laurvick, C., de Klerk, N., Bower, C., Christodolou, J., Ravine, D., Ellaway, C., et al. (2006). Rett syndrome in Australia: A review of the epidemiology. *Journal of Pediatrics, 148*, 347–352.
Lavås, J., Slotte, A., Jochym-Nygren, M., Van Doorn, J., & Witt-Engerström, I. (2006). Communication and eating proficiency in 125 females with Rett syndrome: The Swedish Rett Center survey. *Disability & Rehabilitation, 28*, 1267–1279.
Leonard, H., & Bower, C. (1998). Is the girl with Rett syndrome normal at birth? *Developmental Medicine and Child Neurology, 40*, 115–121.
Luyster, R., Lopez, K., & Lord, C. (2007). Characterizing communicative development in children referred for autism spectrum disorders using the MacArthur-Bates Communicative Development Inventory (CDI). *Journal of Child Language, 34*, 623–654.
Maestro, S., Muratori, F., Barbieri, F., Casella, C., Cattaneo, V., Cavallaro, M. C., et al. (2001). Early behavioral development in autistic children: The first 2 years of life through home movies. *Psychopathology, 34*, 147–152.
Marschik, P. B., & Einspieler, C. (2011). Methodological note: Video analysis of the early development of Rett syndrome – one method for many disciplines. *Developmental Neurorehabilitation, 14*, 355–357.
Marschik, P. B., Einspieler, C., Garzarolli, B., & Prechtl, H. F. (2007). Events at early development: Are they associated with early word production and neurodevelopmental abilities at the preschool age? *Early Human Development, 83*, 107–114.
Marschik, P. B., Einspieler, C., Oberle, A., Laccone, F., & Prechtl, H. F. (2009). Case report: Retracing atypical development: A preserved speech variant of Rett syndrome. *Journal of Autism and Developmental Disorders, 39*, 958–961.
Marschik, P. B., Einspieler, C., Prechtl, H. F., Oberle, A., & Laccone, F. (2010). Relabelling the preserved speech variant of Rett syndrome. *Developmental Medicine and Child Neurology, 52*, 218.
Marschik, P. B., Einspieler, C., & Sigafoos, J. (2012). Contributing to the early detection of Rett syndrome: The potential role of auditory Gestalt perception. *Research in Developmental Disabilities, 33*, 461–466.
Marschik, P. B., Kaufmann, W. E., Einspieler, C., Bartl-Pokorny, K. D., Wolin, T., Pini, G., et al. Profiling early socio-communicative development in five young girls with the preserved speech variant of Rett syndrome. *Research in Developmental Disabilities*, http://dx.doi.org/10.1016/j.ridd.2012.04.012, in press.
Marschik, P. B., Lanator, I., Freilinger, M., Prechtl, H. F., & Einspieler, C. (2011). Early signs and later neurophysiological correlates of Rett syndrome. *Klinische Neurophysiologie, 42*, 22–26.
Marschik, P. B., Pini, G., Bartl-Pokorny, K. D., Duckworth, M., Gugatschka, M., Vollmann, R., et al. (2012). Early speech-language development in females with Rett syndrome: Focusing on the preserved speech variant. *Developmental Medicine and Child Neurology, 54*, 451–456.
Matson, J. L., Fodstad, J. C., & Boisjoli, J. A. (2008). Nosology and diagnosis of Rett syndrome. *Research in Autism Spectrum Disorders, 2*, 601–611.
Matson, J. L., Wilkins, J., & González, M. (2008). Early identification and diagnosis of autism spectrum disorders in young children and infants: How early is too early? *Research in Autism Spectrum Disorders, 2*, 75–84.
Neul, J. L., Kaufmann, W. E., Glaze, D. G., Christodolou, J., Clarke, A. J., Bahi-Buisson, N., et al. (2010). Rett syndrome: Revised diagnostic criteria and nomenclature. *Annals of Neurology, 68*, 944–950.
Nomura, Y., & Segawa, M. (1990). Clinical features of the early stage of the Rett syndrome. *Brain and Development, 12*, 16–19.
Ozonoff, S., Iosif, A., Young, M., Hepburn, G. S., Thompson, S., Colombi, M. C., et al. (2011). Onset patterns in autism: Correspondence between home video and parent report. *Journal of American Academy of Child and Adolescent Psychiatry, 50*, 796–806.
Palomo, R., Belinchón, M., & Ozonoff, S. (2006). Autism and family home movies: A comprehensive review. *Developmental and Behavioral Pediatrics, 27*, S59–S68.
Rowe, M. L., & Goldin-Meadow, S. (2009). Early gesture selectively predicts later language learning. *Developmental Science, 12*, 182–187.
Rowe, M. L., Özçalışkan, Ş , & Goldin-Meadow, S. (2008). Learning words by hand: Gesture's role in predicting vocabulary development. *First Language, 28*, 182–199.
Saint-Georges, C., Cassel, R. S., Cohen, D., Chetouani, M., Laznik, M.-C., Maestro, S., et al. (2010). What studies of family home movies can teach us about autistic infants: A literature review. *Research in Autism Spectrum Disorders, 4*, 355–366.
Sauer, E., Levine, S. C., & Goldin-Meadow, S. (2010). Early gesture predicts language delay in children with pre- or perinatal brain lesions. *Child Development, 81*, 528–539.
Shumway, S., & Wetherby, A. M. (2009). Communicative acts of children with autism spectrum disorders in the second year of life. *Journal of Speech, Language and Hearing Research, 52*, 1139–1156.
Sigafoos, J., Arthur-Kelly, M., & Butterfield, N. (2006). *Enhancing everyday communication for children with disabilities*. Baltimore: Brookes Publishing Company.
Sigafoos, J., Woodyatt, G., Keen, D., Tait, K., Tucker, M., Roberts-Pennell, D., et al. (2000). Identifying potential communicative acts in children with developmental and physical disabilities. *Communication Disorders Quarterly, 21*, 77–86.
Sigafoos, J., Woodyatt, G., Tucker, M., Roberts-Pennell, D., & Pittendreigh, N. (2000). Assessment of potential communicative acts in three individuals with Rett syndrome. *Journal of Developmental and Physical Disabilities, 12*, 203–216.
Stone, W. L., Ousley, O. Y., Yoder, P. J., Hogan, K. L., & Hepburn, S. L. (1997). Nonverbal communication in two- and three-year-old children with autism. *Journal of Autism and Developmental Disorders, 27*, 677–696.

Tams-Little, S., & Holdgrafer, G. (1996). Early communication development in children with Rett syndrome. *Brain and Development, 18*, 376–378.
Thal, D., & Tobias, S. (1992). Communicative gestures in children with delayed onset of oral expressive vocabulary. *Journal of Speech and Hearing Research, 35*, 1281–1289.
Tomasello, M., Striano, T., & Rochat, P. (1999). Do young children use objects as symbols? *British Journal of Developmental Psychology, 17*, 563–584.
Trevarthen, C., & Hubley, P. (1978). Secondary intersubjectivity: Confidence, confiding, and acts of meaning in the first year. In A. Lock (Ed.), *Action, gestures, and symbol* (pp. 183–229). New York: Academic Press.
Vallotton, C. D. (2009). Do infants influence their quality of care? Infants' communicative gestures predict caregivers' responsiveness. *Infant Behaviour & Development, 32*, 351–365.
Volterra, V., & Erting, C. J. (1990). *From gesture to language in hearing and deaf children*. New York: Springer.
von Tetzchner, S. (1997). Communication skills among females with Rett syndrome. *European Child & Adolescent Psychiatry, 6*, S33–S37.
Wetherby, A. M., Yonclas, D. G., & Bryan, A. A. (1989). Communicative profiles of preschool children with handicaps: Implications for early identification. *Journal of Speech and Hearing Disorders, 54*, 148–158.
Woodyatt, G. C., & Ozanne, A. E. (1992a). Communication abilities and Rett syndrome. *Journal of Autism and Developmental Disorders, 22*, 155–173.
Woodyatt, G. C., & Ozanne, A. E. (1992b). Communication abilities in a case of Rett syndrome. *Journal of Intellectual Disability Research, 36*, 83–92.
Woodyatt, G. C., & Ozanne, A. E. (1993). A longitudinal study of cognitive skills and communication behaviours in children with Rett syndrome. *Journal of Intellectual Disability Research, 37*, 419–435.
Yoder, P. J., Warren, S. F., Kim, K., & Gazdag, G. (1994). Facilitating prelinguistic communication in very young children with developmental disabilities. II. Systematic replication and extensions. *Journal of Speech and Hearing Research, 37*, 193–204.
Zampini, L., & D'Odorico, L. (2009). Communicative gestures and vocabulary development in 36-month-old children with Down's syndrome. *International Journal of Language & Communication Disorders, 44*, 1063–1073.